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EMA/COMP/10141/2015
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Recombinant human aspartylglucosaminidase for the treatment of aspartylglucosaminuria

On 15 January 2015, orphan designation (EU/3/14/1410) was granted by the European Commission to ACE Biosciences A/S, Denmark, for recombinant human aspartylglucosaminidase for the treatment of aspartylglucosaminuria.

What is aspartylglucosaminuria?

Aspartylglucosaminuria is an inherited disease belonging to the larger family of metabolic disorders called 'lysosomal storage diseases'. Patients with this condition lack the enzyme aspartylglucosaminidase, which is involved in the breakdown of molecules called glycoproteins within cells. As a result, glycoproteins accumulate in tissues, causing cell damage, particularly in the nerves and bones.

Patients with the disease have developmental delays (such as in speech and movement) in the early years of life, followed by a gradual decline in mental abilities in later years. Patients may also have problems with their bones, such as abnormalities in the curvature of their spine and in their facial features.

Aspartylglucosaminuria is a debilitating and life-threatening disease because of the decline in mental functions and other problems such as epilepsy and infections.

What is the estimated number of patients affected by the condition?

At the time of designation, aspartylglucosaminuria affected not more than 1 in 10,000 people in the European Union (EU). This was equivalent to a total of not more than 51,000 people*, and is below the ceiling for orphan designation, which is 5 people in 10,000. This is based on the information provided by the sponsor and the knowledge of the Committee for Orphan Medicinal Products (COMP).

*Disclaimer: For the purpose of the designation, the number of patients affected by the condition is estimated and assessed on the basis of data from the European Union (EU 28), Norway, Iceland and Liechtenstein. This represents a population of 511,100,000 (Eurostat 2014).

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What treatments are available?

No satisfactory methods for the treating aspartylglucosaminuria were authorised in the EU at the time of designation.

How is this medicine expected to work?

This medicine contains a copy of the enzyme aspartylglucosaminidase that is lacking in patients with aspartylglucosaminuria. When given by injection to the patient, the medicine is expected to replace the missing enzyme and start breaking down the accumulated glycoproteins. This is expected to relieve the symptoms of the disease.

The enzyme in this medicine is made by a method known as 'recombinant DNA technology': it is made by cells into which a gene (DNA) has been introduced that makes them able to produce the enzyme.

What is the stage of development of this medicine?

At the time of submission of the application for orphan designation, the evaluation of the effects of the medicine in experimental models was ongoing.

At the time of submission, no clinical trials with the medicine in patients with aspartylglucosaminuria had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for aspartylglucosaminuria or designated as an orphan medicinal product elsewhere for this condition.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 11 December 2014 recommending the granting of this designation.

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Opinions on orphan medicinal product designations are based on the following three criteria:

- the seriousness of the condition;
- the existence of alternative methods of diagnosis, prevention or treatment;
- either the rarity of the condition (affecting not more than 5 in 10,000 people in the EU) or insufficient returns on investment.

Designated orphan medicinal products are products that are still under investigation and are considered for orphan designation on the basis of potential activity. An orphan designation is not a marketing authorisation. As a consequence, demonstration of quality, safety and efficacy is necessary before a product can be granted a marketing authorisation.

For more information

Sponsor's contact details:

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For contact details of patients' organisations whose activities are targeted at rare diseases see:

- Orphanet, a database containing information on rare diseases, which includes a directory of patients' organisations registered in Europe;
- European Organisation for Rare Diseases (EURORDIS), a non-governmental alliance of patient organisations and individuals active in the field of rare diseases.

Translations of the active ingredient and indication in all official EU languages[1], Norwegian and Icelandic

Language	Active ingredient	Indication
English	Recombinant human aspartylglucosaminidase	Treatment of aspartylglucosaminuria
Bulgarian	Рекомбинантна човешка аспартилглюкозаминидаза	Лечение на аспартилглюкозаминурия
Croatian	Rekombinantna ljudska aspartilglukozaminidaza	Liječenje aspartilglukozaminurije
Czech	Rekombinantní humánní aspartylglukosaminidáza	Léčba aspartylglukosaminurie
Danish	Rekombinant human aspartylglucosaminidase	Behandling af aspartylglukosaminuri
Dutch	Recombinant humaan aspartylglucosaminidase	Behandeling van aspartylglycosaminurie
Estonian	Rekombinantne inimese aspartüülglükosaminidaas	Aspartüülglükosaminuuria ravi
Finnish	Rekombinantti ihmisen aspartyyliglukosaminidaasi	Aspartyyliglukosaminurian hoito
French	Aspartylglucosaminidase humaine recombinante	Traitement de l'aspartylglucosaminurie
German	Rekombinante humane Aspartylglukosaminidase	Behandlung von Aspartylglukosaminurie
Greek	Ανασυνδυασμένη ανθρώπινη ασπαρτυλγλυκοσαμινιδάση	Θεραπεία της ασπαρτυλγλυκοσαμινουρίας
Hungarian	Rekombináns humán aszpartil-glükózaminidáz	Aspartylglucosaminuria kezelése
Italian	Aspartilglucosaminidasi ricombinante umana	Trattamento dell'aspartilglucosaminuria
Latvian	Rekombinantā cilvēka aspartilglikozaminidāze	Aspartilglikozaminūrijas ārstēšana
Lithuanian	Rekombinantinė žmogaus aspartilgliukozaminidazė	Aspartilgliukozaminurijos gydymas
Maltese	Aspartylglucosaminidase uman rikombinanti	Kura tal-aspartilglukosaminurja
Polish	Rekombinowana ludzka asparaginyloglukozoaminidaza	Leczenie asparaginyloglukozoaminurii
Portuguese	Aspartilglicosaminidase humana recombinante	Tratamento da aspartilglicosaminúria
Romanian	Aspartilglucozaminidază umană recombinantă	Tratamentul aspartilglucozaminuriei
Slovak	Rekombinantná humánna aspartylglukozaminidáza	Liečba aspartylglukozaminúrie

[1] At the time of designation

Language	Active ingredient	Indication
Slovenian	Rekombinantna humana aspartilglukozaminidaza	Zdravljenje aspartilglukozaminurije
Spanish	Aspartilglucosaminidasa recombinante humana	Tratamiento de la aspartilglucosaminuria
Swedish	Rekombinant humant aspartylglukosaminidas	Behandling av aspartylglukosaminuri
Norwegian	Rekombinant human aspartylglukosaminidase	Behandling av aspartylglukosaminuri
Icelandic	Raðbrigða manna aspartýlglúkósamínídasi	Meðferð við aspartýlglýkósamínúríu